



## Editorial letter

## Informed consent and patient registry for the rare disease community: Editorial

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Informed consent is a fundamental ethical requirement in most clinical research. Much has been written about the benefits and barriers of research-related informed consent from patients prior to participating in medical research studies. Consent templates and examples exist and are widely available for use by organizations or individual investigators. Appropriately designed templates provide the required elements as mandated by federal regulations and international standards. Nonetheless, research consent forms are often long and complex and not always useful to participant understanding.

In order to initiate, conduct and complete meaningful studies on rare diseases in an appropriate time frame, it is necessary to identify and locate patients with rare diseases who may be dispersed over large geographical areas. Patient registries are becoming an important tool and resource to accelerate patient recruitment into clinical trials and other medical studies designed to evaluate diagnostics and therapeutics with the hope of eventually effectively treating or curing the millions of people suffering from rare diseases.

As more and more patient registries are being developed by patient advocacy groups, research investigators, industry, and other organizations, they are faced with the challenge of the appropriate process for obtaining consent from patients to include their medical histories or donated biospecimens in the patient registries. Importantly, obtaining consent for including patient information and specimens in a registry can be different than obtaining consent for clinical trials or other medical studies that registry patients might ultimately be invited to enroll in. Whether and how extensively federal regulations, such as those found in the Common Rule or HIPAA apply to the establishment of a patient registry will depend on the context in which medical information and

specimens are collected, how they will be stored, and other related factors.

The literature and guidance regarding obtaining consent for participation in patient registries are evolving. Organizations and patient advocacy groups involved in the process of establishing patient registries may be faced with uncertainties regarding the necessary elements and content of informed consent documents. One of the challenges is ensuring that participants receive the information they need to make an informed decision about participating in the registry.

Although the information provided and the requirements for obtaining consent from participants when establishing a patient registry may not be as stringent as consent for participating in a clinical research study, efforts should be made to develop a consent process that adequately informs patients about the purpose for joining the registry, how the registry will be used, and how the confidentiality of their information will be protected.

There is an urgent need in the rare disease community for further discussions among the various stake holders about the process of obtaining consent for participation in a patient registry. Stake holders should work together to develop sets of recommendations for informed consent language and processes that can be adopted by the many different developers of patient registries. As is true for consent forms for individual clinical research studies, informed consent for participating in a patient registry is dependent upon the intended uses for data gathered and analyzed in the patient registry. One consent form cannot fit all the different registries being developed by different entities.

In addition, the consent form is only part of the process of consent. For many rare diseases, adequate information may not exist and may need to be developed and provided to patients (and their family members as appropriate) as part of an ongoing consent process. Providing additional information beyond what can be presented in a written consent form may be essential in order to obtain meaningful consent from participants. The rare diseases community should work together to address these issues and develop templates and recommendations for obtaining the informed consent of patients, facilitating participation in patient registries, and adequately protecting the information provided.

Guidance and recommendations regarding points to consider and information to include in developing a consent process and the consent form would be very helpful to rare disease patient advocacy organizations and others when establishing patient registries. In addition, development and utilization of common templates would facilitate global harmonization necessary to exchanging, sharing, and aggregating de-identified patient medical information from patients around the world for future studies which is so desired by the rare disease community.

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